

CLAIMS

We claim:

1. A method of detecting spinocerebellar ataxia type 10 in a sample containing DNA from an individual to be tested comprising the step of measuring the presence or absence of DNA expansion at a gene locus associated with spinocerebellar ataxia type 10.

2. The method of claim 1, wherein the expansion is measured by the steps of:
extracting the DNA from a sample to be tested;
amplifying the extracted DNA; and
identifying the presence or absence of a DNA expansion in the amplified extension products.

3. The method of claim 1, wherein the sample to be tested is selected from the group consisting of blood, semen, saliva, sweat, urine, nipple aspirates, vaginal swabs, tissue, or a combination thereof.

4. The method of claim 1, wherein the amplification is by PCR.

5. The method of claim 1, wherein the primers are of the sequence of SEQ ID NO: 3 and SEQ ID NO: 4.

6. The method of claim 1, wherein the DNA expansion is measured by Southern blotting analysis of restriction enzyme digests with a probe to the *SCA10* locus.

7. The method of claim 6, wherein the restriction endonuclease is selected from the group consisting of EcoRI, EcoRV, HindIII and BglI.

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8. The method of claim 6, wherein the probe is created by the use of primers of the sequence of SEQ ID NO: 6 and SEQ ID NO: 7.
9. The method of claim 1, where the DNA expansion is determined by pulsed field gel electrophoresis.
10. The method of claim 1, where the DNA expansion is determined by fluorescence *in situ* hybridization.
11. The method of claim 1, where the DNA expansion is comprised of a pentanucleotide repeat.
12. The method of claim 1, where the pentanucleotide repeat is ATTCT.
13. The method of claim 12, wherein the pentanucleotide repeat is repeated between 10 and 29 times for unaffected individuals and greater than 800 times for individuals affected with spinocerebellar ataxia type 10, with individuals having pentanucleotide repeats in between the normal and expanded range requiring additional study for a diagnosis of spinocerebellar ataxia type 10.
14. A method of detecting pentanucleotide repeats in *SCA10* comprising the steps of:
 - isolating DNA from an individual to be tested; and
 - performing PCR analysis using the primers of the sequence of SEQ ID NO: 3 and SEQ ID NO: 4.
15. The method of claim 14 wherein the pentanucleotide repeat is ATTCT.
16. A method of diagnosing spinocerebellar ataxia type 10 comprising the steps of:
 - isolating DNA from an individual to be tested;

performing PCR analysis using the primers of the sequence of SEQ ID NO: 3 and SEQ ID NO: 4;

assessing the number of ATTCT repeats based on comparison to DNA from an unaffected individual; and

determining whether the number of ATTCT repeats is expanded in comparison to that of unaffected individuals.

17. A method of diagnosing spinocerebellar ataxia type 10 comprising the steps of:

isolating DNA from an individual to be tested;

performing PCR analysis using the primers of the sequence of SEQ ID NO: 10 and SEQ ID NO: 11; and

assessing whether the number of ATTCT repeats is expanded in comparison to that of unaffected individuals.

18. A kit for diagnosis of spinocerebellar ataxia type 10 comprising primers suitable for amplifying *SCA10* and an enzyme for amplifying nucleic acids.

19. The kit of claim 18 wherein the primers are the sequence of SEQ ID NO: 3 and SEQ ID NO: 4.

20. The kit of claim 18 wherein the primers are the sequence of SEQ ID NO: 10 and SEQ ID NO: 11.

21. A kit for diagnosis of spinocerebellar ataxia type 10 comprising a probe capable of hybridizing to *SCA10* and an enzyme for amplifying nucleic acids.

22. The kit of claim 21 wherein the probe is created using primers of the sequences of SEQ ID NO: 6 and SEQ ID NO: 7.

23. A non-human transgenic eukaryote wherein the eukaryote is not expressing SCA10; or the ortholog thereof.

24. The non-human transgenic eukaryote of claim 23, wherein said eukaryote is a mammal.

25. The non-human transgenic eukaryote of claim 23, wherein said eukaryote is a mouse.

26. A non-human transgenic eukaryote that is over-expressing SCA10; or the ortholog thereof as compared to a similar non-transgenic eukaryote.

27. The non-human transgenic eukaryote of claim 26, wherein said eukaryote is a mammal.

28. The non-human transgenic eukaryote of claim 26, wherein said eukaryote is a mouse.